EUROPEAN DNA PROFILING GROUP (EDNAP) MEETING

DUBLIN - 2 - 3 SEPTEMBER 2004

Host: Maureen Smyth & Geraldine O'Donnell
Chairman: Niels Morling

A list of participants is attached as Annex 1.

1. Welcome
Geraldine O'Donnell welcomed members to Dublin.

2. Update on publications

2.1 Y chromosome SNPs – Santiago II — Angel Carracedo
The paper is accessible at Forensic Sci Int online publications:
Brion M, Dupuy BM, Heinrich M, Hohoff C, Hoste B, Ludes B, Mevag B, Morling N, Niederstätter, Parson W, Sanchez J, Bender K, Siebert N, Thacker C, Vide MV, Carracedo A. A collaborative study of the EDNAP group regarding Y-chromosome binary polymorphism analysis. Forensic Sci Int 2004, in press.

3. Update on EDNAP exercises

3.1 mtDNA from head hair samples – II — Niels Morling
Seven labs submitted results. Innsbruck performed further typings. Presently, there are 115 mtDNA sequences of 41 hairs. Discordant results will be scrutinized if the original electropherogrammes can be recovered. A batch of hairs that were sent from Rome to Innsbruck gave typing results that were highly unlikely if the hairs came from the same person. Therefore, these results will be excluded from the analysis.

3.2 EMPOP — Walther Parson
Walther Parson gave an update on the EMPOP project. The internet site has been remodelled and software has been modernized (Search Engine V 2.0). There is an EMPOP test site (URL, account and password will be communicated), which includes a small number (87) of data from 7 countries (Austria, Finland, Germany, Japan, Kenya, Morocco, and Russia) representing the 3 subcontinents (Africa, East-Asia, West-Eurasia). The data consist of HV1-, HV1/HV2- and CR-sequences in order to reflect the complexity of mtDNA data available in the individual datasets.
Action
EDNAP laboratories are asked to evaluate the EMPOP test site. It is useful and necessary that feedback is made on the performance of the database including the usability of the site, the correctness of the plausibility checks, and the correctness of the output of the search programme.

An annotated table of the 87 sequences included in the EMPOP test site will be provided in a separate file. Please respond to Walther Parson by October 15th. Earlier comments (as soon as they become apparent) are highly appreciated.

3.3 Telogen hair STR typing — Hermann Schmitter

Herman Schmitter reported that the method is now being offered by a number of laboratories in Germany and repeated his invitation to help with the method.

4. Updates from other groups

5.1 ENFSI

ENFSI has invited EDNAP representatives to meet the ENFSI philosophy group in order to discuss and coordinate work.

5.2 SNPforID — Peter Schneider

A total of 52 unlinked, autosomal SNP markers have been selected to form a core SNP set. Non-binary SNPs exhibiting 3 alleles, population-specific SNPs and SNPs with skewed allele frequency distributions have been identified. A set of 29 Y-chromosomal SNPs that identifies major Y haplogroups has been selected and a SNP multiplex created.

The Institute of Legal Medicine in Innsbruck has been affiliated to the SNPforID consortium, and work has been initiated in order to characterize non-coding mtDNA SNPs to further characterize common haplogroups.

Two autosomal multiplexes - a 23-plex and a 29-plex – have been developed and combined into a single 52-plex that can be typed by two SNaPshot minisequencing reactions. Studies using MALDI TOF MS, Nanogen electronic microarrays and spotted SBE-TAG arrays are continued.

"Whole Genome Amplification" with the GenomiPhi kit (Amersham) gave good results using 1 ng of high molecular weight DNA with up to a 2,000 fold yield of amplified genomic DNA. Drop-out and drop-in artefacts were observed in STR and SNP typing of amplificates of partly degraded DNA. Thus, GenomiPhi can be used to preserve DNA fragments of e.g. precious, non-degraded DNA samples, but it cannot be recommended for degraded DNA samples.

The SNPforID consortium is bound by the EU contract to protect the intellectual property rights on knowledge produced. New knowledge will be protected by material transfer agreements for an interim period until the information has been published and, thereby, made generally available in the public domain.

5.3 SWGDAM — Peter Gill

A PowerPoint presentation from Samuel Baechtel is attached as Annex 2.

5.4 NIST — Niels Morling

A PowerPoint presentation from John Butler is attached as Annex 3.

6. Commercial DNA investigations of ancestry and physical characteristics — All members

Commercial companies claim to be able to provide DNA investigations that e.g. can tell the ancestry or the colour of the eyes of a person. Such genetic markers have been known for years. Examples: Some combinations of autosomal STR/VNTR types are associated with ethnicity as well as eye, hair and skin colour, some Y-chromosome haplogroups/haplotypes are associated with geographic origin, certain autosomal SNPs are associated with colour of

hair and skin and possibly also eye colour. However, most of the presently known associations between genetic factors and physical traits are weak.

7. Future activities

7.1 Standardisation of STRs in Europe Peter Gill

The need for future changes in the genetic markers in crime DNA databases was discussed. Presently, Interpol recommends a core set of 7 STRs. With the growing number of individuals in the European DNA databases, a need for an increased discriminatory power of the core set is anticipated. It was decided that EDNAP will work for a scientifically sound long term solution that includes clarification of the goals and the possible ways forward. It was recognised that (1) flexibility of the databases must be created by introduction of rational strategies and relevant software support, (2) the chemistry must be adjusted to allow the generation of larger multiplexes, and (3) new genetic systems must be selected according to rational criteria, including the relevance of the new systems in crime case work. Peter Gill will circulate a first draft of a discussion document (alternatively two documents considering (1) strategies for expansion of databases and (2) strategies for selection of new genetic systems).

7.1 New exercises

7.1.1 SNP-STR exercise Peter Gill

Peter Gill will – in collaboration with John Butler - organise a comparative exercise on the performance of

- Conventional STR typing
- Low Copy Number STR typing
- Typing with short STRs from NIST
- Typing with the autosomal SNP package from the FSS (21 SNPs with amplicons < 150 bp, LCN sensitivity, fully validated and ready for publication).

Six stains with heavily degraded DNA will be sent to 10 participating laboratories.

7.1.2 mtDNA SNP exercise Walther Parson/Angel Carracedo

Walther Parson will – in collaboration with colleagues in Santiago – organise an exercise on mtDNA-SNP typing. The panel of SNPs is to be decided. Reagents as well as control samples will be sent to participating laboratories. The participating laboratories shall perform a population study.

7.2 Interpretation of results of DNA mixtures, LCN, etc. Peter Gill

The need for recommendations on interpretation of results of DNA mixtures, LCN, etc. was recognised by members. Peter Gill felt that consensus can be obtained between scientists in the US and Europe. The members encouraged the board of the ISFG to activate the DNA Commission of the ISFG. Peter Gill will write to the board of the ISFG concerning the problems to be dealt with and propose a ‘road map’ for the activities of the DNA Commission.

7.3 Joint meetings with ENFSI

Members were in favour of organising the next EDNAP meeting in connection to the next ENFSI meeting.

8. EDNAP website update Peter Schneider

URL of the web site: www.isfg.org/ednap/ednap.htm.
Peter Schneider will circulate instructions and password for the member site by E-mail.
Please contact Peter Schneider if you have suggestions for the EDNAP web site.

9. Next meeting

The next EDNAP meeting will be held in conjunction with the next ENFSI meeting in early 2005. The date and place is to be decided.

10. Any other business

There was no other business and the meeting closed with sincere thanks to Maureen Smyth and Geraldine O'Donnell.